DOCUMENT RESUME

ED 148 072 EC 103 455

TITLE Seminar on Usher's Syndrome: Proceedings.
INSTITUTION Rochester School for the Deaf, N.Y.
PUB DATE 76
NOTE 34p.; Held at Rochester School for the Deaf (Rochester, New York, December 14, 1976)
AVAILABLE FROM Rochester School for the Deaf, 1545 St. Paul Street, Rochester, New York 14621

EDRS PRICE MF-$0.83 HC-$2.06 Plus Postage.
DESCRIPTORS Clinical Diagnosis; Conference Reports; *Deaf Blind; *Etiology; *Genetics; Incidence; Information Dissemination; Medical Evaluation; Multiply Handicapped; Prevention; Rehabilitation
IDENTIFIERS *Ushers Syndrome

ABSTRACT

Summarized are the presentation of M. Vernon and the Comments of primary panelists from a seminar on Usher's Syndrome, a genetic disease involving congenital deafness and progressive loss of vision due to retinitis pigmentosa. The following topics are addressed: genetics today, nature of Usher's Syndrome, symptoms, prevalence, lay diagnosis for referral, medical diagnosis, treatment, prevention, public information, available rehabilitation and other services, and research. Following panelists' comments and audience questions is a bibliography of studies and reports on the topic of Usher's Syndrome. Appended are a sample parent letter and a screening form from the Rochester School for the Deaf. (SBH)

****
* Documents acquired by ERIC include many informal unpublished *
* materials not available from other sources. ERIC makes every effort *
* to obtain the best copy available. Nevertheless, items of marginal *
* reproducibility are often encountered and this affects the quality *
* of the microfiche and hardcopy reproductions ERIC makes available *
* via the ERIC Document Reproduction Service (EDRS). EDRS is not *
* responsible for the quality of the original document. Reproductions *
* supplied by EDRS are the best that can be made from the original. *
****

ED148072

U S DEPARTMENT OF HEALTH, EDUCATION & WELFARE
NATIONAL INSTITUTE OF EDUCATION

THIS DOCUMENT HAS BEEN REPRODUCED EXACTLY AS RECEIVED FROM THE PERSON OR ORGANIZATION ORIGINATING IT. POINTS OF VIEW OR OPINIONS STATED DO NOT NECESSARILY REPRESENT OFFICIAL NATIONAL INSTITUTE OF EDUCATION POSITION OR POLICY

SEMINAR ON

USHER'S SYNDROME

Sponsored by:
Rochester School for the Deaf
and
the RSD Parent/Teachers' Association

"PERMISSION TO REPRODUCE THIS MATERIAL HAS BEEN GRANTED BY

The Rochester School for the Deaf

TO THE EDUCATIONAL RESOURCES INFORMATION CENTER (FRIC) AND USERS OF THE ERIC SYSTEM."

EC103455

PROCEEDINGS OF A SPECIAL SEMINAR ON USHER'S SYNDROME

HELD AT THE ROCHESTER SCHOOL FOR THE DEAF

FOREWORD

Out of our concern and in response to numerous requests to the Rochester School for the Deaf (RSD) for information regarding Usher's Syndrome, RSD organized a seminar which dealt with this topic. The seminar was held on Tuesday, December 14, 1976, on the RSD campus.

The format of the program was a presentation by Psychologist McCay Vernon, Ph.D., from Western Maryland College, followed by a panel discussion in which seven panelists related their experiences with and knowledge of Usher's Syndrome in relation to deafness and retinitis pigmentosa.

This document encompasses a summary of Dr. Vernon's presentation as well as the comments of the primary panelists in response to questions from the audience. Following these summaries is a suggested bibliography summarizing studies or reports on the topic of Usher's Syndrome.

We wish to express our appreciation to all of the seminar participants for their contributions to the program. A special note of appreciation goes to: the Parent/Teachers' Association of RSD for its support in sponsoring this conference; to Shirley R. Curtis, Director of RSD Student Personnel Services, who was the major thrust behind the organization of this seminar; and to Gail Carlson for her recording of the conference.

Leonard G. Zwick
Superintendent

TABLE OF CONTENTS

Page

. Introduction of Panelists 1

. Presentation by Dr. McCay Vernon 2
"Usher's Syndrome: A Genetic Disease Involving Congenital Deafness and Progressive Loss of Vision Due to Retinitis Pigmentosa"

Genetics Today 2

Nature of Usher's Syndrome 4

Symptoms 4

Prevalence 6

Lay Diagnosis for Referral 6

Medical Diagnosis 7

Treatment 10

Prevention 10

Public Information 11

Available Rehabilitation and Other Services . . .12

Research 15

Summary 16

References 17

. Panelists' Comments 18

. Audience Questions 21

. Suggested Bibliography 25

. Appendices 28

INTRODUCTION OF PANELISTS

The seminar commenced with Superintendent of the Rochester School for the Deaf, Leonard G. Zwick, welcoming parents, staff, and interested parties. Mr. Zwick commented that the purpose of the meeting was twofold: to discuss accurate information on Usher's Syndrome and to acquaint persons with the possible courses of action for person's having Usher's Syndrome.

Mr. Zwick then turned the program over to Marsden Fox, M.D., (Pediatrician, RSD Medical Director, and Clinical Assistant Professor of Pediatrics, University of Rochester Medical School) who introduced and moderated the resource panel. The panel, whose purpose was to react both to the speaker and the audience, consisted of:

- McCay Vernon, Ph.D., Psychologist, Western Maryland College, Guest Speaker
- John Fitzgerald, M.D., Ophthamologist, Clinical Assistant Professor, University of Rochester Medical School
- John Kiley, M.D., Child Psychiatrist, Consultant to RSD, Attending Child Psychiatrist, Rochester Mental Health Center, Clinical Instructor of Psychiatry, University of Rochester Medical School
- Phillip L. Townes, M.D., Professor of Anatomy, Genetics, Pediatrics, University of Rochester Medical School
- Donald Sims, Ph.D., Audiologist, Research Assistant in Audiology, National Technical Institute for the Deaf
- Leonard G. Zwick, M.A., M.Ed., Educator, Superintendent of Rochester School for the Deaf
- Vicki Hurwitz, Student, Deaf Individual, Parent, Homemaker
- There were other resource persons in the audience including Barbara Martin, regional representative from the Helen Keller National Center for Deaf/Blind Youths and Adults.

Following is a summary of Dr. McCay Vernon's presentation.

Usher's Syndrome: A Genetic Disease Involving Congenital Deafness and Progressive Loss of Vision Due to Retinitis Pigmentosa

McCay Vernon, Ph.D.*

Usher's Syndrome is a relatively rare disease in the general population. However, among those who are deaf and hard of hearing it is all too common. In a general sense Usher's characterizes one of the major diagnostic problems medicine faces today. To clarify this requires some background information on genetics, of which Usher's is a case in point.

Genetics Today

Genetics is currently causing a revolution in medicine comparable to Pasteur's discovery of a germ theory of disease in the 1800's.[10, 11] For example, today 25 percent of infant mortality is due to congenital defects, most of which are genetic. By contrast in 1900 only 3.3 percent of those deaths were congenital. Antibiotics, vaccines, and other medical advances have eliminated or reduced the effects of many infectious diseases that used to kill and maim.[10, 11]

---

*Reprint request to Dept. Psychology, Western Maryland College, Westminster, Maryland 21157

This change poses a unique, extremely difficult challenge for the physician. In the past a doctor by becoming familiar with a relatively few major infectious disease such as diphtheria, measles, scarlet fever, and polio could diagnose the medical problems of most or at least many of his patients.

By contrast, today with the increasing importance of genetic disease the situation is reversed. There are now 2,000 known hereditary disorders with more being discovered all the time.[10, 11] Even the most common of these is extremely rare. Many are disorders a physician may only see once or twice in a lifetime. For example, cystic fibrosis is perhaps the most common of severe terminal genetic diseases. Yet it occurs in only one per 2,000 people. Other genetic disorders affect only certain ethnic groups. For instance, Tay Sachs (severe mental retardation, general disintegration and early death) occurs primarily in Eastern European Jews. Sickle cell anemia, a fatal blood disorder, strikes primarily Blacks.

Understandably there is often a failure on the part of the physician to diagnose such relatively rare disorders. The results can be tragic. For example, Usher's Syndrome, which should be diagnosed early in life, is usually not identified until adulthood.[9] Consequently patients may waste years in inappropriate professional preparation. They may unknowingly expose themselves to dangers of death because they are unaware of their visual defects. In many respects Usher's Syndrome epitomizes the serious problem of providing diagnosis and treatment for genetic disorders.

Nature of Usher's Syndrome

Usher's Syndrome is an inherited condition. It results in the double handicap of hearing loss and a progressive loss of vision (due to retinitis pigmentosa) which eventually leads to legal blindness. In a few cases of Usher's there are other neurophysiological sequelae such as mental retardation, loss of vestibular function, and epilepsy.[7, 9] The hearing loss is usually congenital, severe, and stable. It should be noted that retinitis pigmentosa can also occur without hearing loss.[4]

Symptoms

Nightblindness:

The first symptom to appear in the course of the disease is nightblindness. It is usually present fairly early in childhood but rarely diagnosed. At first the victim does not realize that his night vision is defective. He thinks everybody sees as he does. Thus, he blames his clumsiness for the falls and bumps he experiences. One individual related that as a youngster he noticed how much better other children get around at night while playing hide and seek. He accounted for this by assuming they had superior memories. He thought that during the day they noticed where things were. Consequently, at night they were able to avoid the objects he was continually running into.

As night blindness progresses it becomes an increasing hazard often leading to automobile accidents and serious falls.[6] It also isolates the individual because going out at night alone becomes impossible. Even when accompanied by

a friend it tends not be very satisfying because dark restaurants, clubs, and other dimly lit places prevent the person with Usher's from communicating in sign language or otherwise seeing what is going on.

Narrowing of Visual Field:

Usually during the teen-age years the visual field starts to narrow. However, this "tunneling of vision" is rarely diagnosed until the patient is in his twenties or sometimes thirties. As in the case of the night blindness, the loss of peripheral vision is due primarily to rod function deteriorating.[2]

Because diagnosis is usually not made until late, persons with Usher's Syndrome frequently continue to drive until their vision field is down to 15 to 20 degrees. This is in sharp contrast to the normal visual field of 180 degrees. Thirty-eight states measure only central visual acuity in driver's license exams, thereby overlooking cases of "tunnel vision." Hence, legally blind people with Usher's Syndrome pass the driving test and represent automobile hazards to themselves and others.

Legal Blindness:

Often by the late twenties or thirties the visual field is so narrowed that the individual is legally blind. However, central visual acuity may remain adequate for reading up until the fifties or sixties. However, usually between age 25 to age 45 the individual is forced to give up his job due to his failing eyesight.[7]

It is important to note that there is great variation in the progression of visual loss. In rare cases legal blindness is present as early as six years of age. Others retain considerable useful vision up into their fifties or sixties.[7]

Prevalence

The prevalence data on Usher's Syndrome are somewhat tenuous. The following statistics reflect the best available information.[7] About 3 per 100,000 people in the U.S. are victims. Of all those who have a congenital hearing impairment, three to six percent are also victims of Usher's Syndrome. In certain areas such as parts of Louisiana where consanquinious marriages are common the figure may run to 30 percent.

Another way to look at prevalence is to note that there are 70 to 100,000 people in the United States who have retinitis pigmentosa.[2] Of these, one out of four has Usher's Syndrome. Thus, there are 17,500 to 25,000 people in this country who will be deaf-blind due to Usher's Syndrome. One person per hundred is a carrier of the gene for the disease.[7]

The major point of all of these somewhat conflicting statistics is that Usher's Syndrome and the relatively frightening condition of deaf-blindness which it involves are all too common among deaf people. Unfortunately, professionals such as ophthalmologists, family practitioners, teachers, optometrists, audiologists, otolaryngologists, pediatricians, and others who see deaf children have in the past been relatively naive about the disease.[10] This must change. Schools for deaf children, professionals, parents, and deaf people need to know about the problem.

Lay Diagnosis for Referral

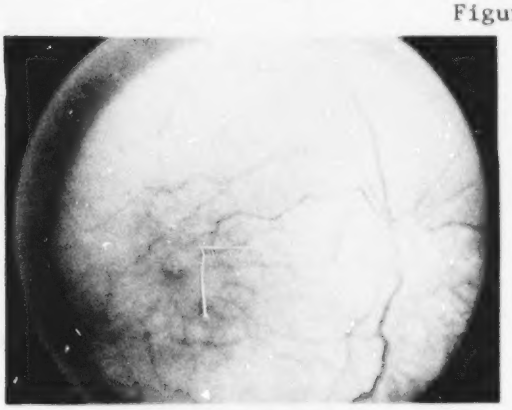
For those who are not ophthalmologists the major symptoms that should lead to an immediate medical referral are nightblindness (sometimes accompanied by a balance problem) and/or tunnel vision.

If those who were around deaf children were aware of these two symptoms they could identify for referral almost all cases of Usher's Syndrome while the patients were in their teens or even before.

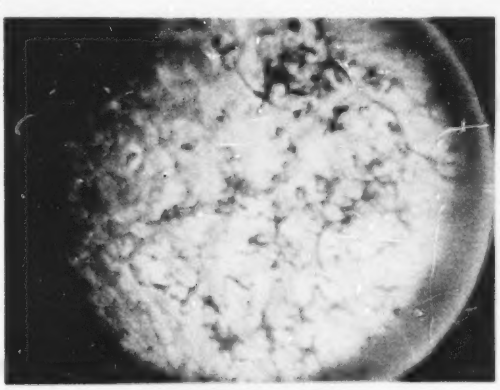
Medical Diagnosis

In the early stages the diagnoses can be very difficult. The patient may not know he has symptoms because he spends most of his time in a well lighted environment and moves his eyes back and forth so that he compensates for deficiences in his visual field. Patients frequently come for routine eye examinations and report no symptoms at all. It is not uncommon for people with Usher's Syndrome to pass regular eye examinations at ages well into their twenties. The ophthamologist or the family doctor may look into the eye and actually see what appears to be a normal fundus even though the patient may have the early stages of the disease. By adolescence the fundus examination may show the beginnings of vessel narrowing in the retina and some pigment migration around the edges of the retina.

Figure I



Vessel Narrowing



Pigment Migration

Electroretinography:

Because the disease is difficult to detect on a routine eye examination some ophthalmologists feel that patients should have a specialized test procedure called electroretinography.[2] In brief this procedure involves the patient looking at a flashing light while wearing a contact lens which is electrically wired to measure the retina responses. The impulses coming out of the eye tell the ophthalmologist whether or not the patient has the early stages of Usher's Syndrome.[2,] For patients who are at risk for Usher's Syndrome, Dr. Berson of the Massachusetts Eye and Ear Infirmary recommends testing between the ages of 6 and 15. At that age the recording can be done without any anesthesia and the diagnoses can be made with certainty.[2, 3] Diagnosis with electroretinography is also possible during infancy. At this age the patient must be given sedation. Infant testing should be done primarily in very high risk cases where issues of family planning are to be decided. Note that a family which has one child with Usher's Syndrome has a 25 percent chance that each additional child will have the condition.

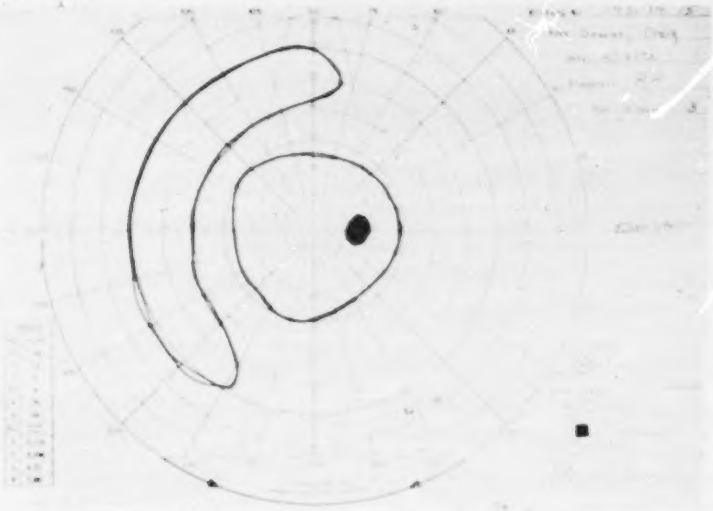
Other Diagnostic Procedures:

Electroretinography, while the most definitive of all diagnostic procedures and by far the best for early diagnosis, is a process which is done only in large medical centers. Thus, in most settings three other tests are used:

Ophthalmic Examination - As Usher's Syndrome progresses the blood vessels of the retina narrow and the black particles of pigment are evident on the retina (Figure I). Because this process occurs first in the periphery of the retina it is important that the pupils be dilated for ophthalmoscopy. When dilated the periphery of the retina can be seen better and diagnosis made earlier.[7, 10]

Dark Adaptation Testing - This procedure measures the lack of sensitivity to light which is one of the basic symptoms of Usher's Syndrome. The test requires extensive communication between physician and patient. Thus, if it is to be done validly with a deaf patient there should be a sign language interpreter present.

Visual Field Testing - The normal person's visual field is about 180°. As the second major symptom of Usher's Syndrome is a narrowing of the visual field, the disease can be diagnosed by measuring a patient's loss of peripheral vision (Figure II).



Narrowing of Visual Field

Treatment

At present there is no known treatment for Usher's Syndrome.[7, 22] Many approaches have been tried including vitamin A therapy, tissue transplants, enzyme injections shots, etc. None have demonstrated success. The advances which have been made are more in the area of rehabilitation. Mobility training, and prosthetic developments such as the nightscope are examples.

Prevention

Because Usher's Syndrome is incurable and involves the tragic double handicap of deafness and blindness, primary effort should be devoted to prevention and to the rehabilitation of those who already have Usher's Syndrome. The major approach to prevention is through visual screening and genetic counseling. The deaf are the high risk population for Usher's Syndrome. During school years and to a certain extent in later life deaf people are a highly centralized available population. The yield on a visual screening of the deaf would be around three percent and would go up to six percent if only the genetically deaf were screened.[7] Bergsma has developed a pilot approach that is practical and relatively inexpensive.[8] The cost would be infinitesimal compared to the cost of educating, training, or even merely providing custodial care for a deaf-blind person. To educate one deaf-blind child for 13 years costs $182,000, enough to pay for several years of nationwide screening of the high risk population.[7]

If screening were combined with followup genetic counseling for patients with Usher's and their parents and relatives, there would be a significant reduction in the prevalence of Usher's. However, the condition cannot be eradicated by this means, because it is transmitted by an autosomal recessive gene. This means men and women may be carriers even though they do not have Usher's Syndrome. Recessive genes are not expressed unless the same gene is present on both members of a pair of chromosomes. Thus, recessive genes produce an effect only when they are transmitted to the newborn by both parents. Autosomal genes are those not carried on a sex chromosome. Because the gene causing Usher's is autosomal, both men and women can have Usher's.

Screening and genetic counseling pose many ethical and philosophical problems, which is one reason Bergsma advocates only a pilot program initially.[1] However, when the possible drawbacks of counseling are weighed against the awesome nature of deaf-blindness, it is apparent that they are the lesser evil.

Public Information

Until physicians, optometrists, and other professions who work with deaf children know about Usher's Syndrome, diagnoses will continue to be made ten or twenty years later then they should. Tragic errors in family planning and career choice will result. Research, screening, genetic counseling, prevention, and rehabilitation are all depending upon public and professional awareness. After years of total neglect embryonic efforts in this direction are now being made. For example, the National Foundation March of Dimes has circulated an article on Usher's Syndrome to

geneticists.[3, 10] Gallaudet College has held two symposia on the disease, published proceedings from these, and now has a film available on Usher's Syndrome. The Southwest Regional Deaf-Blind Center has hosted a workshop and published proceedings. The Retinitis Pigmentosa Foundation is doing the most through its television spots, and its National Registry of persons with retinitis pigmentosa and its circulation of informative printed materials.

Much more must be done. For example, if ophthalmologists knew about Usher's Syndrome they would routinely check all deaf patients by at least examining the periphery of the retina with an ophthalmoscope, preferably after dilating the pupil of the eye. Also, visual field testing would be done more often, and referrals would be made for dark adaptation testing and electroretinography.

Available Rehabilitation and Other Services

It is helpful to propose solutions to problems surrounding Usher's Syndrome. But what is available now for people with Usher's, or for that matter for other deaf-blind people? Resources include:

A. Helen Keller National Center for Deaf-Blind Youths and Adults (111 Middle Neck Road, Sands Point, New York 11050). The National Center offers a diagnosis, evaluation, training, and follow up for any deaf-blind person meeting its admission requirements. It also has regional representatives throughout the United States. The National Center should be one of the first places contacted by deaf-blind persons and their families.

B. Bureau of Education for the Handicapped (Services for Deaf-Blind Children, Department of Health, Education, and Welfare, 400 Maryland Ave., S.W., Washington, D.C. 20202). The Bureau controls ten Regional Centers for Deaf-Blind Children that provide primarily diagnosis and evaluation. Anyone knowing of a deaf-blind child should notify the Bureau which will have the nearest Regional Center contact the child and the family.

C. Rehabilitation Services Administration (Department of Health, Education and Welfare, 330 C Street, S.W., Washington, D.C. 20201). This agency administers for federal-state programs of government-supported vocational rehabilitation services for handicapped persons. Rehabilitation counselors are available in almost every community. They can obtain medical, educational and other services for deaf-blind persons who meet certain standards, provided the local rehabilitation budget is adequate.

Ironically, in these times of high unemployment government money to provide job training and education for handicapped people has been cut drastically. Furthermore, most rehabilitation counselors cannot communicate with deaf-blind people and have a limited understanding of their potential, their needs, and what is available for them. Despite these problems if the Rehabilitation Services Administration is contacted they will put the deaf-blind person in contact with a local counselor.

D. Bureau of Supplement Security Income (SSI) (Social Security Administration, 108 West High-Rise, Baltimore, Maryland 21235). This relatively new program offers many deaf-blind people regular subsistence income. Also, enrollment in SSI makes training money that otherwise would be frozen available through the federal-state rehabilitation program. Almost every community has an SSI office.

E. Committee for the Deaf-Blind (American Council of the Blind, 1211 Connecticut Ave., N.W., Suite 506, Washington, D.C. 20036). Formed in 1973, this committee is a Division of the American Council of the Blind and will offer advice and some help to deaf-blind people. Half of its members are themselves deaf-blind.

F. National Retinitis Pigmentosa (RP) Foundation (8331 Mindale Circle, Baltimore, Maryland 21207). The RP Foundation funds research for retinitis pigmentosa and hopes to come up with a cure. It also keeps a national registry of people with Usher's Syndrome, publishing information on devices to help deaf-blind people, and reports on the latest research findings on retinitis pigmentosa.

g. Office of Public Service Programs (Gallaudet College, Kendall Green, Washington, D.C. 20002). They have sponsored two symposiums on Usher's Syndrome and published information about the disease. The program is new. Thus far their primary service has been in the area of public information about Usher's Syndrome and helping college students with the disease.

H. National Association of the Deaf (NAD) (814 Thayer Avenue, Silver Spring, Maryland 20910). The association provides some literature on Usher's Syndrome and has formed a section of its organization to serve on deaf-blind people. A small but significant percent of the membership has Usher's Syndrome. As the world's largest organization for the deaf, NAD has better contact than any other group with Usher's Syndrome people.

I. International Association of Parents of the Deaf (814 Thayer Avenue, Silver Spring, Maryland 20910). This group recently established a committee for parents of deaf-blind children. Services are only beginning, but the committee could become a major asset for parents who have just discovered their child has Usher's Syndrome or is for some other reason deaf-blind.

Research

Although there is at present no treatment for Usher's Syndrome intensive research is underway in an effort to discover a cure or cause of the condition and of other retinal diseases. At the Berman-Gund Laboratory of Massachusetts Eye and Ear Infirmary they are studying the biochemistry of rods and cones because these cells are involved in genetic diseases of the retina.[2, 3] Diet, especially the effects of vitamins and amino acid on retinal function are also

receiving extensive study. For example, the chemical taruine has been found to be related to retina degeneration in studies with cats.[5]

Other research at the Berman-Gund Laboratory involves seeing whether or not protecting the eyes of those with Usher's Syndrome from light help preserve vision a little longer.[4] This is being done in a carefully controlled study using a contact lens that protects one eye from light while the other eye is used for reading and traveling.

Summary

Usher's Syndrome is a recessively inherited condition involving profound congenital hearing impairment and progressive loss of vision leading to blindness. At present it is incurable but prevention efforts and rehabilitation services can reduce its spread and its tragic consequences.

The major symptoms of nightblindness and narrowing visual field are often overlooked by the patient and physician until the condition is advanced. Thus, diagnosis is late with serious consequences for the patient, his family and society. Increased professional and public awareness of Usher's Syndrome is needed to correct the problem.

Present research efforts funded primarily by the Retinitis Pigmentosa Foundation and conducted at the Berman-Gund Laboratory of Massachusetts Eye and Ear Infirmary gives hope of eventual discovery of a cure or a way to arrest the development of the disease. However, at present there is no treatment. Early diagnosis and effective rehabilitation are currently the best services available.

References

1. Bergsma, D.R. Ophthalmic aspects of Usher's Syndrome. Proceedings of Symposium on Usher's Syndrome: Washington, D.C.: Gallaudet Press, 1973, p. 12-18.

2. Berson, Eliot L., M.D. Retinitis Pigmentosa and allied retinal diseases: Electrophysiologic findings. Transactions of the American Academy of Ophthalmology and Otology, 81, 1976, 659-666.

3. Berson, Eliot L., M.D. Hereditary and nutritionally induced retinal degeneration: Electrophysiologic and biochemical studies, Birth Defects, Original Article Series, Vol. XII, 3, 359-372, 1976.

4. Berson, Eliot L., M.D. Light deprivation for early Retinitis Pigmentosa, Arc. of Oph., 1971, 85, 521-529.

5. Schmidt, Berson, and Hayes, K.C. Retinal degeneration in the taurine-deficient cat. Transactions of the American Academy of Ophthalmology and Otology, 81, 1976, 687-693.

6. Vernon, M. Psychological aspects of Usher's Syndrome (In Press 1977). Proceedings of Workshop on Usher's Syndrome. New York: Helen Keller National Center for Deaf-Blind Youths and Adults (111 Middle Neck Road, Sands Point, N.Y.), 1977.

7. Vernon, M. Usher's Syndrome: Problems and some solutions. Hearing and Speech Action, 1976, 44, 6-13.

8. Vernon, M. Overview of Usher's Syndrome: Congenital deafness and progressive loss of vision. Proceedings of Symposium on Usher's Syndrome: Washington, D.C.: Gallaudet Press, 1973, p. 1-11.

9. Vernon, M. Usher's Syndrome - deafness and progressive blindness. Journ. of Chronic Diseases, 22, 1969, 133-151.

10. No Author. Genetic control - threat or therapy? Medical World News, Vol. 13(18), 1972, p. 45-55.

11. No Author. Genetic counseling: Is it needed too little or too much? Medical World News, Vol. 13, 1972, p. 17-18.

SEMINAR PANELISTS' COMMENTS

Following Dr. Vernon's presentation, a film was shown on retinitis pigmentosa, the visual part of the illness of Usher's Syndrome. The meeting was then opened to questions and comments from the audience and the panelists.

Dr. Fitzgerald commented that all the professionals involved with Usher's Syndrome patients are making the public more aware of the disease so they can make earlier diagnoses so patients can be directed vocationally. By an early diagnosis, they can then guide these individuals so they can continue to be productive as long as they are visually active.

Dr. Fitzgerald then went into some of the symptoms of the disease. Usually, the condition of retinitis pigmentosa is chronic, is slowly progressive, and may sometimes have remissions. Some of the sight may return and there may be improvements. At these times, professionals become confused by treatment. All they may be treating are the particular remissions of the disease, and it is not a truly effective therapy. The condition is slow and it may take 10 to 30 years to progress. Therefore, it does not mean that because a person has Usher's Syndrome or retinitis pigmentosa that they will be blind in a matter of weeks or months. One of the first symptoms of the disease, nightblindness, becomes noticeable not in the dark, but in dusk. (For diagnostic purposes for nightblindness, there is an electroretinogram at St. Mary's Hospital in Rochester, New York.) Another symptom, tunnel vision, is like looking down a gun barrel. This condition takes place after all of the cells in the retina have been destroyed. This is the end result, not the beginning, of the disease. This is a progressive disease for

which there is no known treatment. Therefore, we must be stimulated for early diagnosis.

Looking at this from an educational standpoint, Mr. Zwick asked how many places are really looking at the magnitude of the eye problems of hearing-impaired children. To Zwick's knowledge, a review of the literature reveals that there have been five programs in the country which have looked at this through visual screening. These programs include: The Pennsylvania School's Speech and Hearing Center; The Kendall School; California School at Berkeley; St. Mary's School in Buffalo, New York; and the Ontario School. Wherever they have looked among deaf people, they have found tremendous eye problems. At St. Mary's, they have made several diagnoses in the past four - five years of retinitis pigmentosa, and there are three at the school now out of 260 students.

Dr. Sims commented that the Audiology Department at the National Technical Institute for the Deaf has undertaken visual screening of its students. Approximately 10 percent could not meet visual skills requirements using the Bausch and Lomb visual screening tests. NTID has two confirmed cases of retinitis pigmentosa. Audiologically, there are some promising attempts being made to determine the carriers of retinitis pigmentosa and Usher's Syndrome by examining the cells audiometrically. They have found minimum hearing losses among the carriers at certain frequencies. Also, a promising area for future research might be in the area of electrophysiological audiometric techniques.

Dr. Kiley presented a question to Dr. Vernon regarding the anxiety that this syndrome produces in all of us and the subsequent psychiatric symptoms that it produces in people who have Usher's Syndrome and the worries which it brings about in the family. About fifteen years

ago, there were sensory deprivation experiments conducted in which people were isolated visually, tactily, and auditorily. Virtually anyone who participated in the experiment developed psychotic symptoms. In reading some of Dr. Vernon's works, Dr. Kiley was impressed that many of the symptoms ascribed to Usher's Syndrome are very similar to those described in this experiment. Dr. Vernon stated that he didn't know the answer to this question. However, a high percentage of people in the early studies with Usher's Syndrome were diagnosed to be psychotic and retarded. Were these cases of misdiagnosis? Were these reactions to the trauma of being deaf-blind? Or were they part of the same neurophysiological lesion or problem that caused the deafness or blindness.

Dr. McCay Vernon (center) discusses the percentage of deaf people with Usher's Syndrome with Vicki Hurwitz (left) and Jean Dickson (right).

Dr. John Kiley (center), RSD psychiatric consultant, discusses Usher's Syndrome with Dr. Don Sims, audiologist NTID (left), and Chris Evans, audiologist RSD (right).

AUDIENCE QUESTIONS

In answer to an audience question about the black spots in the retina and how they develop, Dr Fitzgerald stated that in this disease, there is a degeneration or deterioration of the rods in the retina. The rods carry light sensation that hits the peripheral portion of the retina. The rods make up 85 - 90 percent of the retina. The cones occupy the central part of the retina and carry all detail vision. Retinitis pigmentosa takes place in the rods. As they deteriorate or degenerate, the side vision narrows down. Associated with this deterioration is a disturbance in the pigmentation cells. The pigmentation grows and spreads from its normal position in relation to the rods out into the retina, particularly around the blood vessels, and assumes a star-shaped form.

Another question was posed about whether there are any other types of retinitis pigmentosa. Dr. Fitzgerald commented that there are many types. The one type being discussed in relation to Usher's Syndrome is probably the most common. Instead of having a black corpuscular condition, there may be a condition where the retina is filled with a white substance scattered throughout the retina. Most cases occur bilaterally, but some occur in just one eye. There are variations and other types of symptoms. When talking about this condition in general, all of the retinitis pigmentosa patients do not go blind because there may be only a segment or a part of the retina that is involved. There may be an abortion of the disease. It may start and stop. Some may progress gradually. It is not just one isolated disease.

A question was asked about which of the various diagnostic tests is the most efficient for mass screening. Dr. Fitzgerald stated that the best way to pick it up is through opthamological examination where the pupils are fully dilated. The first signs you see are the narrowing of the arterials. The electroretinogram is not a device to be used for screening. The narrowing of the arterials can be picked up from the age of three and up. The pigmentation changes happen later in life.

If people suspect they are carriers, can they be tested before they have children? Dr. Townes stated that screening and testing large populations is very difficult. The answer to this question would depend on who is being discussed. If speaking of a brother or sister of a person with Usher's Syndrome or retinitis pigmentosa, this individual has a 50 percent chance of being a carrier and should be examined through very extensive studies for evidence of whether or not he/she is a carrier of the disease. Great efforts should be made when the individual who by blood relationship has a high risk of being a carrier.

From a genetic point of view, Dr. Townes provided a perspective for consideration. Dr. Vernon indicated the great importance of looking at the deaf population in terms of having Usher's Syndrome. Going a step beyond that from a genetic point of view, there are some 2,000 genetic diseases. Usher's Syndrome, rare though it may be, is found to the degree of approximately three - six percent in the hereditarially or genetically deaf population. However, one cannot lose track of the other 50 percent of the deaf population. It is important to look at this other 50 percent to reassure them in terms of future family planning

that their deafness is not hereditary and that they do not have much of a chance of having a child who will be deaf.

In regard to the students at the Rochester School for the Deaf, it was asked how feasible it would be to screen 200 students for retinitis pigmentosa. Dr. Fitzgerald stated that he was sure cooperation could be obtained, and certain students could be selected to be seen in a reasonable length of time. The cost of an examination is approximately $20 and would take from 1-1/2 to 2 hours. If field studies or other tests had to be done, there would be additional time, cost, and effort involved.

Regarding the kinds of jobs that Usher's Syndrome patients can assume and the kinds of rehabilitation programs available to them, Barbara Martin of the Helen Keller Institute commented that it must be kept in mind that the job should not require vision or hearing. At the Helen Keller National Center, a number of deaf-blind people have been placed in assembly-type jobs. Some have returned to their positions as teacher aides, rehabilitation counselors, etc.

In regard to communication and socialization with a person with Usher's Syndrome, Dr. Vernon commented that the first part of deaf-blindness is the terrible isolation. Research has been done on deaf-blindness, and it was found that for eleven hours per day, a deaf-blind person sits with no communication with anyone. A deaf-blind person in a crowd is totally isolated if he/she is more than three feet away from someone. Persons should make an effort to go up to the blind-deaf person and find out how to communicate--fingerspelling or signing

in the hand, writing out block letters in the hand, or any way. Sometimes these people are afraid of human contact, but it is necessary to make an effort. Organizations within the deaf community can communicate with deaf-blind people, provide transportation to deaf clubs and social events, transmit information about the Helen Keller National Center, collect games and books in braille, etc. This could be the topic for another whole discussion.

Dr. Marsden Fox (left), RSD medical director and Leonard G. Zwick, superintendent RSD, discuss future courses of action.

SUGGESTED BIBLIOGRAPHY

The following additional information is available concerning Usher's Syndrome and retinitis pigmentosa.

Anon. "Sightless and Soundless World," Rehab. Rec. 9:24-25 (Mar. - Apr.) 1968.

Akabane, N. "Hearing Impairment in Retinitis Pigmentosa," Jap. J. Ophthal. 12 (3):139 - 146, 1968.

Barrett, Steve, "Visual Screening of the Deaf in the School Setting" South Central Region, Helen Keller National Center for Deaf-Blind Youths and Adults.

Berson, E.L.: Hereditary and nutritionally induced retinal degenerations: Electrophysiological and biochemical studies. The Eye and the Inborn Errors of Metabolism (National Foundation March of Dimes Birth Defect Series, Vol. 12 No. 3). ed. D. Bergsma, A. Bron and E. Cotlier. A. R. Liss. New York, 1976, pp. 359-372.

Berson, E.L.: Retinitis pigmentosa and allied retinal diseases: Electrophysiological findings. Trans. Am. Acad. Ophthalmol. Otolaryngol. 81:659-666, 1976.

Berson, E.L.: Night blindness: Some aspects of management. Clinical Low Vision, ed. E. Gaye, Little Brown and Company, Boston, 1976. pp. 301-306.

Berson, E.L.: Hereditary retinal diseases: Classification with the full-field electroretinogram. Documents Ophthalmologica Proceedings Series No. 12-XIVth ISCERG Symposium, May 10-14, 1976. Louisville, Kentucky, U.S.A., ed. T. Lawwil, Dr. W. Junk. The Hague, The Netherlands, 1977, in press.

Berson, E.L.: Electroretinographic testing as an aid in determining visual prognosis in families with hereditary retinal degenerations. In: Retina Congress, eds. R.C. Pruett and C.D.J. Regan, Appleton-Century-Crofts, New York, 1974, pp. 41-54.

Berson, E.L., Mehaffey, L., and Rabin, A.R.: A night vision pocketscope for patients with retinitis pigmentosa: Design consideration. Arch. Ophthalmol. 91:495-500, 1974.

Bettica, Louis J. "The Deaf-Blind Individual With Usher's Syndrome, A Total Rehabilitation Approach". Proceedings from Workshop on Usher's Syndrome, Long Island, New York, December 1976.

Bettica, Louis J., "Counseling the Usher's Syndrome Deaf-Blind Individual" Presented at the Annual Conference of Professional Rehabilitation Workers with the Adult Deaf, Salt Lake City, Utah, July 1976.

Bettica, Louis J., Prause, Robert J. "Types of Jobs Performed by Deaf-Blind People" Publication of the Helen Keller National Center for Deaf-Blind Youths and Adults, Long Island, New York, 1976.

Curtis, Scott, Donlon, Edward T., and Wagner, Elizabeth (eds.) Deaf-Blind Children: A Program for Evaluating Their Multiple Handicaps. New York: A.F.B., 1970.

Finnegan, Joseph P. Jr., Davis, Walter S., "Usher's Syndrome Workshop" The Florida School Herald, Vol. 76, No. 7., St. Augustine, Florida, March 1977.

Hurst, L.A. "Hereditary Disorders of Childhood and Adolescence: Preventive Action," Med. Proc. 14 (13): 271-281 (June 29) 1968.

Juurmaa, J. "A Comparative Analysis of the Effects of Blindness and Deafness on Psychic Functions," Eye Ear Nose Throat Monthly, 47 (11): 553-561 (Nov.) 1968.

Kloepfer, H.W.; Laguaite, J.K., and McLaurin, J.W. "The Hereditary Syndrome of Congenital Deafness and Retinitis Pigmentosa (Usher's Syndrome)," Laryngoscope 76:850-862, 1966.

Lawson, L.J., Jr.: Myklebust, H.R. "Ophthalmological Deficiencies in Deaf Children" Exceptional Children, September, 1970.

Prause, Robert J., "Placement of Individuals With Usher's Syndrome" Present at the Workshop on Usher's Syndrome, at Helen Keller National Center for Deaf-Blind Youths and Adults, Sands Point, New York, December 1976.

Rosenfeld, Jill., "Communication Techniques with the Deaf-Blind in a Rehabilitation Setting", Presented at the Virginia Chapter of the American Association of Workers for the Blind, Charlottesville, Virginia, August 1975.

Spainer, John, "Evaluating the Individuals Potential", Helen Keller National Center for Deaf-Blind Youths and Adults, Long Island, New York.

Vernon, McCay, Psychological Aspects of Usher's Syndrome, Proceedings from Workshop on Usher's Syndrome. Long Island, New York; Helen Keller National Center for Deaf-Blind Youths and Adults (In press 1977).

Vernon, McCay, Usher's Syndrome: Problems and Some Solutions. Hearing and Speech Action, 44, 1976, 6-13.

Vernon, McCay; Schwartz, Katherine, Usher's Syndrome: A Disease That Causes Some Deaf People to Lose Their Sight. Deaf American, 1974, 26, 3-5.

Vernon, McCay, Overview of Usher's Syndrome: Congenital Deafness and Progressive Loss of Vision. Volta Review, 76, 1974, 100-105.

Vernon, McCay, Overview of Usher's Syndrome: Congenital Deafness and Progressive Loss of Vision Proceedings of Symposium on Usher's Syndrome. Gallaudet College Press: Wash., D.C., 1974, 1-11.

Vernon, McCay, Multiple Handicapped Deaf Children: Current Status in Readings in Exceptional Children. New York: Appleton, Century and Crofts, Psychology Series (2nd Edition) 1972, 379-396. Edited by Trapp, E.P. and Himelstein, P.)

Vernon, McCay, Usher's Syndrome--Deafness and Progressive Blindness: Clinical Cases, Prevention, and Literature Survey. Journal of Chronic Diseases, 22, 1969, 133-151. Reprinted for Circulation to geneticists and physicians by the Jonas Salk Biological Institute, The March of Dimes, and The Birth Injuries Foundation, 1971. Also reprinted in the Rehabilitation Teacher, Jan. and Feb. 1971. VIII #1, 3-12, #2, 1-16.

Vernon, McCay, Usher's Syndrome--Deafness and Progressive Blindness: An Abstract. The New Outlook for the Blind, 64, 1970, 51-52.

Vernon, McCay, Usher's Syndrome--A Premium on Prevention. Sight Saving Review, 39, 1969, 152.

Films

"The World of Deaf-Blind Children - How they Communicate" 16mm Color/ Sound, 29 min., Free Loan Basis, Film Librarian, Guidance Information Center, Cory Hill, Saxtons River, Vermont 05154.

"The World of Deaf-Blind Children-Growing Up" 16mm Color/Sound, 29 min.) Free Loan Basis, Film Librarian, Guidance Information Center, Cory Hill, Saxtons River, Vermont 050154.

"Retinitis Pigmentosa" 16 mm, Color/Sound, National Retinitis Pigmentosa Foundation, Rolling Park Building, 8331 Mindale Circle, Baltimore, Maryland 21207.

Appendix I

ROCHESTER SCHOOL FOR THE DEAF
1545 St. Paul Street
Rochester, New York 14621
716-544-1240

Dear Parents,

As you know, your child's vision is extremely important since he/she has a severe/profound hearing impairment. There are many diseases that can affect your child's vision as well as problems of acuity; we are asking that all of our students be seen by an ophthalmologist (eye physician). This year we are asking your ophthalmologist to give special attention to screening for retinitis pigmentosa which when coupled with deafness is called Usher's Syndrome. This particular syndrome often leads to total blindness in addition to the deafness and early diagnosis is essential.

Please make an appointment with your ophthalmologist (M.D. eye doctor) as soon as possible and have him complete the enclosed form.

If you need help in securing an appointment, please contact our school nurse who will be happy to help.

Thank you for your prompt attention to this matter.

Sincerely,

Shirley R. Curtis CSW
Director, Student Personnel
Services

Leonard G. Zwick
Superintendent

Appendix II

ROCHESTER SCHOOL FOR THE DEAF
1545 St. Paul Street
Rochester, New York 14621
716-544-1240

Dear Examiner:

We ask that you give special attention to screening for retinitis pigmentosa because when it is coupled with deafness (Usher's Syndrome) the problems it presents to the individual and his educational/vocational program are multiplied. An early diagnosis is essential for good planning. It has been found that field vision screening is effective in older patients and the electroretinogram (ERG) and the electrooculargram (EOG) will permit early definite diagnosis.

Thank you.

Marsden Fox M.D.
Medical Director

PLEASE GIVE SPECIAL ATTENTION TO SCREENING FOR RETINITIS PIGMENTOSA

EYE EXAMINATION RECORD

_____
Pupil's Name

Entered RSD. _____ Birth Date _____

Cause of Deafness: _____

DATES OF EXAMINATIONS: _____

History -
Comments

Vision:									
Without Correction	:R	L	B	:R	L	B	:R	L	B
With Correction	:R	L	B	:R	L	B	:R	L	B
Present Glasses	:R			:R			:R		
	:L			:L			:L		
	:R			:R			:R		
Retinoscopy	:L			:L			:L		
	:R			:R			:R		
Acceptance	:L			:L			:L		
R	:R			:R			:R		
X	:L			:L			:L		
External	:R			:R			:R		
Examination	:L			:L			:L		
Media &	:R			:R			:R		
Fundi	:L			:L			:L		

Diagnosis

_____

Recommendations

Recommendations for wearing glasses _____
Safety-Shatter-Proof Lens _____
Do you plan to see this child again? If so, when? _____
Are modifications of the school program indicated?
If so, what? _____

Signature of Examiner _____ Title _____

Date _____

PLEASE GIVE SPECIAL ATTENTION TO SCREENING FOR RETINITIS PIGMENTOSA.